

NEWBORN SCREENING PROGRAM  
**ACMG RECOMMENDATIONS**  
APRIL 2005

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The following disorders are included in the ACMG recommendation for a national uniform newborn screening panel.

Currently included in Washington's panel

- 1 Biotinidase deficiency
- 2 Congenital adrenal hyperplasia
- 3 Congenital hypothyroidism
- 4 Galactosemia (GALT)
- Hemoglobinopathies:
  - 5 Sickle cell anemia
  - 6 Hb S/Beta-thalassemia
  - 7 Hb S/C disease
- 8 Homocystinuria
- 9 Maple syrup urine disease (MSUD)
- 10 Medium chain acyl-CoA dehydrogenase (MCAD) deficiency
- 11 Phenylketonuria (PKU)

Under current consideration by the Board

- 12 Hearing loss
- 13 Cystic fibrosis (CF)

Remaining (all are detected through MS/MS screening of dried blood spot)

- 14 3-Methylcrotonyl-CoA carboxylase deficiency
- 15 3-OH 3-CH3 glutaric aciduria
- 16 Argininosuccinic acidemia
- 17 Beta-ketothiolase deficiency
- 18 Carnitine uptake defect
- 19 Citrullinemia
- 20 Glutaric acidemia type I
- 21 Isovaleric acidemia
- 22 Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
- 23 Methylmalonic acidemia
- 24 Methylmalonic acidemia (mutase deficiency)
- 25 Multiple carboxylase deficiency
- 26 Propionic acidemia
- 27 Trifunctional protein deficiency
- 28 Tyrosinemia type I (TYR I)
- 29 Very long-chain acyl-CoA dehydrogenase deficiency

Removed after release of report

- ~~30 Glucose-6-phosphate dehydrogenase (G6PD) deficiency~~